RAB18 gene

RAB18, member RAS oncogene family

Normal Function

The *RAB18* gene provides instructions for producing the RAB18 protein, which functions as a GTPase. Often referred to as molecular switches, GTPases can be turned on and off. They are turned on (active) when they are attached (bound) to a molecule called GTP and are turned off (inactive) when they are bound to another molecule called GDP. When active, RAB18 is involved in a process called vesicle trafficking, which moves proteins and other molecules within cells in sac-like structures called vesicles. RAB18 regulates the movement of substances between compartments in cells and the storage and release of fats (lipids) by structures called lipid droplets. The protein also appears to play a role in a process called autophagy, which helps clear unneeded materials from cells. RAB18 is important for the organization of a cell structure called the endoplasmic reticulum, which is involved in protein processing and transport.

Health Conditions Related to Genetic Changes

RAB18 deficiency

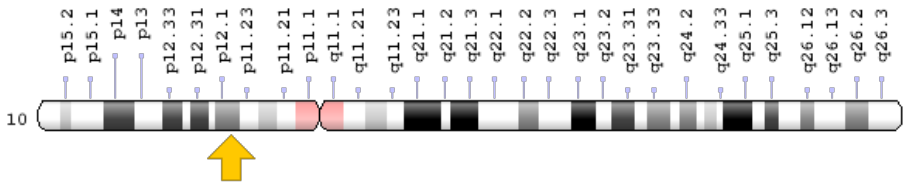
At least five mutations in the *RAB18* gene have been found to cause Warburg micro syndrome, which is the most severe of the disorders caused by RAB18 deficiency. Warburg micro syndrome is characterized by multiple eye abnormalities, vision impairment, severe intellectual disability, and a reduction of the hormones that direct sexual development (hypogonadotropic hypogonadism).

The *RAB18* gene mutations that cause Warburg micro syndrome eliminate the function of the RAB18 protein. It is unclear how a shortage (deficiency) of RAB18 activity leads to eye problems, brain abnormalities, and other features of Warburg micro syndrome.

Chromosomal Location

Cytogenetic Location: 10p12.1, which is the short (p) arm of chromosome 10 at position 12.1

Molecular Location: base pairs 27,504,304 to 27,542,239 on chromosome 10 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- RAB18 small GTPase
- RAB18LI1
- ras-related protein Rab-18 isoform 1
- ras-related protein Rab-18 isoform 2
- ras-related protein Rab-18 isoform 3
- ras-related protein Rab-18 isoform 4
- ras-related protein Rab-18 isoform 5
- WARBM3

Additional Information & Resources

Educational Resources

- Basic Neurochemistry: Molecular, Cellular and Medical Aspects (6th edition, 1999): Small G proteins
  https://www.ncbi.nlm.nih.gov/books/NBK28084/

Clinical Information from GeneReviews

- RAB18 Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK475670

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28RAB18%5BTIAB%5D%29+OR+%28RAB18+small+GTPase%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- RAS-ASSOCIATED PROTEIN RAB18
  http://omim.org/entry/602207

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_RAB18.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=RAB18%5Bgene%5D
- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:14244
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:22931
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/22931
- UniProt
  https://www.uniprot.org/uniprot/Q9NP72

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